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Other non-peer reviewed scholarship

Professional educational materials or reports, in print or other media:

Local/Unpublished Clinical Guidelines and Reports:

Thesis:

Chung, W.K. Genetics of obesity in rodents and man [dissertation]. [New York]: Rockefeller University; 1996. 452 p.

Manuscripts Submitted to Preprint Servers

Abstracts, Poster Presentations, and Exhibits Presented at Professional Meetings:

Narrative Report

Introduction

I am a physician scientist specializing in human genetics. After completing my training in June 2002 at Columbia University, I joined the faculty at Columbia University and was promoted through the ranks to my final position in 2017 as the tenured Kennedy Family Professor of Pediatrics in Medicine. In July 2023, I joined Boston Children's Hospital and HMS where I provide administrative leadership to the Department of Pediatrics, provide clinical care, supervise trainees, and conduct research on human genetics of several diseases (autism, neurodevelopmental disorders, KIF1A associated neurological disorder, congenital heart disease, congenital diaphragmatic hernia, esophageal atresia), pulmonary

hypertension, obesity, diabetes, and newborn screening to enhance our knowledge and the quality of patient care. I am a member of the National Academy of Medicine. I am seeking appointment to the rank of Professor with Investigation as my Area of Excellence with Significant Supporting Activities of Clinical Expertise and Administration and Institutional service. I devote 30% of my time to research, 15% to clinical practice, 5% to teaching, and 50% to administration.

Area of Excellence - Investigation

I have been conducting clinical research on human genetic diseases for over 30 years. I have elucidated the genetic basis of many human diseases including contributions from rare de novo genetic variants, inherited rare variants, and common variants and demonstrated associations with clinical outcomes based upon the underlying genetic etiology. I have identified over 50 novel genetic conditions and characterized the clinical phenotype. Three of these conditions bear my name. I have translated advances in genetic diagnostics including the development of multigene panels by clinical indication, implemented exome and genomic sequencing for clinical diagnostics in prenatal and pediatric medicine, and how am assessing the ability to use genomic integrated risk assessment for common conditions in adults and children. I have performed pilot studies for newborn screening for SMA, Duchenne muscular dystrophy, and now am using genome sequencing in GUARDIAN to screen for ~250 genetic conditions simultaneously and have demonstrated synergies with traditional newborns sceening and am working toward population based genomic screening. I have begun to develop treatments for some of these conditions including an ASO strategy currently in clinical trial for selection knock down of a dominant negative heterozygous mutation in KIF1A. I have received funding for my research from several institutes at NIH including NICHD, NHGRI, NHLBI, NIDDK, NINDS, NCI, and private foundations including the Simons Foundation and CZI. All of my research aims to determine the genetic basis for human disease, tailor care based upon genomic information, and implement genomics into medical care in an ethical, cost effective, and equitable manner. I lead many large national and international consortia including DHREAMS, CARE, SPARK, and Simons Searchlight.

Teaching

I have been actively involved in teaching and supervision of medical students, dental students, residents, fellows, postdocs, and junior faculty since I joined the Columbia faculty. I have personally mentored over 100 students in my research laboratory including thesis advisor, scholarly project advisor, thesis committee, and fellowship mentor. I was responsible for designing the course in human genetics for the first year medical and dental students at Columbia and have been the course director for the last 20 years and consistently receive outstanding assessments for the course. I have served as the fellowship director for our Molecular Genetics and Cytogenetics ABMGG training program. I was the Associate Director for Training in our NCI Comprehensive Cancer Center and the Medical Director for our genetic counseling graduate program. I frequently teach in CME courses. I have received numerous awards for teaching including the Presidential Award for Outstanding Teaching, Columbia's highest teaching award, and the Women's mentorship award from the AMA. I have written over 80 major chapters in textbooks and reviews in the literature. I also teach the public and patients through numerous videos online describing our research findings and including major TED talks with over 4 million views.

Significant Supporting Activity - Clinical expertise

My clinical interests and expertise revolve around the practice of genetics. Since 2002 I have been actively involved in clinical care, providing treatment for patients with genetic conditions; I have led

the neurofibromatosis center of excellence, the von Hippel Lindau center of excellence, founded and directed the DISCOVER program for undiagnosed diseases, and the TREATMENT program to develop new treatments. I receive referrals from colleagues locally, regionally, nationally, and internationally. Patients around the world seek me out to diagnose rare undiagnosed conditions. Patients also seek me out for my expertise in clinical conditions I have described. The programs that I created have grown and now evaluate and treat ~1000 patients each year and serve as a core training experience for medical students, residents, and fellows.

Significant Supporting Activity - Administration and Institutional Service

At Columbia, I served as the Director of Clinical Genetics and the Director of Cancer Genetics. I served as the Director of the Precision Medicine Resource of our Irving Institute of Clinical Research. I have served as a national leader in human genetics as a member of council at the National Human Genome Research Institute, the scientific advisory board of All of Us, a member of the board of the American Society of Human Genetics and position as Treasurer elect, organizer for the 2023 Gordon Conference on Human Genetics and Genomics, and as the original plaintiff in the Supreme Court case that overturned gene patents.

Summary

Since my appointment as Chief, Department of Pediatrics at BCH at HMS I have endeavored to provide excellent clinical care, administrative oversight, and teaching, while also conducting research and clinical trials in rare genetic conditions. Through my lecturing (at a regional, national and international level), my written works with an h index of 115, my clinical research, and my involvement with professional societies I have sought to improve the care for patients with genetic conditions I look forward to working on identification of the genetic basis for human disease, characterizing rare genetic conditions, and implementing genomic medicine at scale to improve health.